



Ochoa syndrome

Ochoa syndrome is a disorder characterized by urinary problems and unusual facial expressions.

The urinary problems associated with Ochoa syndrome typically become apparent in early childhood or adolescence. People with this disorder may have difficulty controlling the flow of urine (incontinence), which can lead to bedwetting. Individuals with Ochoa syndrome may be unable to completely empty the bladder, often resulting in vesicoureteral reflux, a condition in which urine backs up into the ducts that normally carry it from each kidney to the bladder (the ureters). Urine may also accumulate in the kidneys (hydronephrosis). Vesicoureteral reflux and hydronephrosis can lead to frequent infections of the urinary tract and kidney inflammation (pyelonephritis), causing damage that may eventually result in kidney failure.

Individuals with Ochoa syndrome also exhibit a characteristic frown-like facial grimace when they try to smile or laugh, often described as inversion of facial expression. While this feature may appear earlier than the urinary tract symptoms, perhaps as early as an infant begins to smile, it is often not brought to medical attention.

Approximately two-thirds of individuals with Ochoa syndrome also experience problems with bowel function, such as constipation, loss of bowel control, or muscle spasms of the anus.

Frequency

Ochoa syndrome is a rare disorder. About 150 cases have been reported in the medical literature.

Genetic Changes

Ochoa syndrome can be caused by mutations in the *HPSE2* gene. This gene provides instructions for making a protein called heparanase 2. The function of this protein is not well understood.

Mutations in the *HPSE2* gene that cause Ochoa syndrome result in changes in the heparanase 2 protein that likely prevent it from functioning. The connection between *HPSE2* gene mutations and the features of Ochoa syndrome are unclear. Because the areas of the brain that control facial expression and urination are in close proximity, some researchers have suggested that the genetic changes may lead to an abnormality in this brain region that may account for the symptoms of Ochoa syndrome. Other researchers believe that a defective heparanase 2 protein may lead to problems with the development of the urinary tract or with muscle function in the face and bladder.

Some people with Ochoa syndrome do not have mutations in the *HPSE2* gene. In these individuals, the cause of the disorder is unknown.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- hydronephrosis-inverted smile
- hydronephrosis with peculiar facial expression
- inverted smile and occult neuropathic bladder
- inverted smile-neurogenic bladder
- partial facial palsy with urinary abnormalities
- UFS
- urofacial Ochoa's syndrome
- urofacial syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Ochoa syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0403555/>

Other Diagnosis and Management Resources

- GeneReview: Urofacial Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK154138>
- National Institute of Diabetes and Digestive and Kidney Diseases: Urodynamic Testing
<https://www.niddk.nih.gov/health-information/diagnostic-tests/urodynamic-testing>
- Scripps Health: Self-Catheterization -- Female
<https://www.scripps.org/articles/3954-self-catheterization-female>
- Scripps Health: Self-Catheterization -- Male
<https://www.scripps.org/articles/4190-self-catheterization-male>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Bilateral Hydronephrosis
<https://medlineplus.gov/ency/article/000474.htm>
- Encyclopedia: Neurogenic Bladder
<https://medlineplus.gov/ency/article/000754.htm>
- Encyclopedia: Reflux Nephropathy
<https://medlineplus.gov/ency/article/000459.htm>
- Health Topic: Urine and Urination
<https://medlineplus.gov/urineandurination.html>

Genetic and Rare Diseases Information Center

- Ochoa syndrome
<https://rarediseases.info.nih.gov/diseases/104/ochoa-syndrome>

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: The Urinary Tract and How It Works
<https://www.niddk.nih.gov/health-information/urologic-diseases/urinary-tract-how-it-works>
- National Institute of Diabetes and Digestive and Kidney Diseases: Urodynamic Testing
<https://www.niddk.nih.gov/health-information/diagnostic-tests/urodynamic-testing>

Educational Resources

- Disease InfoSearch: Ochoa syndrome
<http://www.diseaseinfosearch.org/Ochoa+syndrome/5288>
- MalaCards: urofacial syndrome 1
http://www.malacards.org/card/urofacial_syndrome_1
- Orphanet: Ochoa syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2704

Patient Support and Advocacy Resources

- National Kidney Foundation
<https://www.kidney.org/>
- National Organization for Rare Disorders
<https://rarediseases.org/rare-diseases/urofacial-syndrome/>

GeneReviews

- Urofacial Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK154138>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ochoa+syndrome%5BTIAB%5D%29+OR+%28urofacial+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- UROFACIAL SYNDROME 1
<http://omim.org/entry/236730>

Sources for This Summary

- Al Badr W, Al Bader S, Otto E, Hildebrandt F, Ackley T, Peng W, Xu J, Li J, Owens KM, Bloom D, Innis JW. Exome capture and massively parallel sequencing identifies a novel HPSE2 mutation in a Saudi Arabian child with Ochoa (urofacial) syndrome. *J Pediatr Urol.* 2011 Oct;7(5):569-73. doi: 10.1016/j.jpuro.2011.02.034. Epub 2011 Mar 29.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21450525>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3157539/>
- Aydogdu O, Burgu B, Demirel F, Soygur T, Ozcakar ZB, Yalcinkaya F, Tekgul S. Ochoa syndrome: a spectrum of urofacial syndrome. *Eur J Pediatr.* 2010 Apr;169(4):431-5. doi: 10.1007/s00431-009-1042-9. Epub 2009 Aug 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19669792>

- Daly SB, Urquhart JE, Hilton E, McKenzie EA, Kammerer RA, Lewis M, Kerr B, Stuart H, Donnai D, Long DA, Burgu B, Aydogdu O, Derbent M, Garcia-Minaur S, Reardon W, Gener B, Shalev S, Smith R, Woolf AS, Black GC, Newman WG. Mutations in HPSE2 cause urofacial syndrome. *Am J Hum Genet.* 2010 Jun 11;86(6):963-9. Erratum in: *Am J Hum Genet.* 2010 Aug 13;87(2):309.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20560210>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3032078/>
- Derbent M, Melek E, Arman A, Uçkan S, Baskin E. Urofacial (ochoa) syndrome: can a facial gestalt represent severe voiding dysfunction? *Ren Fail.* 2009;31(7):589-92.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19839856>
- Garcia-Minaur S, Oliver F, Yanez JM, Soriano JR, Quinn F, Reardon W. Three new European cases of urofacial (Ochoa) syndrome. *Clin Dysmorphol.* 2001 Jul;10(3):165-70.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11446407>
- Ochoa B. Can a congenital dysfunctional bladder be diagnosed from a smile? The Ochoa syndrome updated. *Pediatr Nephrol.* 2004 Jan;19(1):6-12. Epub 2003 Nov 25. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/14648341>
- Pang J, Zhang S, Yang P, Hawkins-Lee B, Zhong J, Zhang Y, Ochoa B, Agundez JA, Voelckel MA, Fisher RB, Gu W, Xiong WC, Mei L, She JX, Wang CY. Loss-of-function mutations in HPSE2 cause the autosomal recessive urofacial syndrome. *Am J Hum Genet.* 2010 Jun 11;86(6):957-62. Erratum in: *Am J Hum Genet.* 2010 Jul 9;87(1):161. Fisher, Richard B [added].
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20560209>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3032074/>
- Stamatiou K, Tyrizis S, Karakos C, Skolarikos A. Urofacial syndrome: a subset of neurogenic bladder dysfunction syndromes? *Urology.* 2011 Oct;78(4):911-3. doi: 10.1016/j.urology.2010.12.061. Epub 2011 Apr 13.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21492912>

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